



PROPOSED AMENDED  
FIGURES 4-6

09/707,919

4/18

[http://www.ncbi.nlm.nih.gov/SNP/snp\\_retrieve.cgi?subsnp\\_id=8697](http://www.ncbi.nlm.nih.gov/SNP/snp_retrieve.cgi?subsnp_id=8697)



SNP Details

## SNP Details

### General

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### SNP:

Handle|local\_snp\_id: FGU-CBT | SKB.2K.1.1

NCBI Assay Id(ss#): 869704

Reference SNP Id(rs#): 695871

STS Information: Not submitted

### From SNP Database:

Submitter Handle: FGU-CBT  
Submitter Batch ID: SKB.2K.1  
Release Date: Aug 2 2000 2:53PM  
Molecular type: Genomic  
No. of Chromosomes sampled: 215  
Synonym defined:

Organism: Homo sapiens

Population: INDRP

Submitter Method ID: SCA2-SNP

Citation: Single Nucleotide Polymorphism in SCA2 Gene.

[View citation details](#)

### Search

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[Chromosome Report](#)

NCBI Assay ID: 869704  
Submitter SNP ID: SKB.2K.1.1  
Synonyms:  
LOCUSID: 6311  
Submitter STS ID: not available  
STS Accession: U70323  
GenBank Accession: Human ataxin-2 gene/ Spinocerebellar ataxia 2 (SCA2) gene  
Gene Name: 459  
Length:

### Flanking Sequence Information:

5' Assay: CTCCGCCCTCA GACTGTTTGT GTAGCAACGG CAACGGCGGC GCGCGGTTTC GGCCCGGCTC  
CCGGCGGCTC CTTGGTCTCG GCGGGCCTCC CCGCCCTTC GTGCTC (SEQ ID NO: 14)

Observed: G/C

3' Assay: TCCTTCTCCC CCTCGCCAGC CCGGGCGCCC CTCGGGCGGC GCCAACCCGC GCCTCCCCGC  
TCGGCGCCCG TCGGTCCCG CCGCGTTCG CCGTCTCCTT GGCGCGCCCG GCTCCCGGCT  
GTCCCCGCC GCGGTGGAG CCGGTGTATG GCGCCCTCAC CATGTGCT (SEQ ID NO: 15)

3' Flank: GAAGCCCCAG CAGCAGCAGC AGCAGCAGCA GCAACAGCAG CAGCAGCAAC AGCAGCAGCA  
GCAGCAGCAG CAGCGCGCGC CCGCGGCTGC CAATGTCCGC AAGCCCGCGC GCAGCGGCT  
TCTAGCGTCG CCGCGCGCGC CGCCTTCGCC GTCTCTGTC TCGGTCTCCT CGTCTCGGC  
CAC (SEQ ID NO: 16)

### Allele Frequency Information:

POP Batch Id: SKB.2K.1:

Submitter Handle: FGU-CBT  
Submitter Method ID: SCA2-SNP  
Citation: Single Nucleotide Polymorphism in SCA2 Gene.

Handle|PopulationID: FGU-CBT | INDRP  
No. of Chromosomes Sampled: 215

Details

Allele: C = 0.293 / G = 0.707

Fig.4(Cont.)

9/26/00 6:50 PM



Get SNP Record

# Reference SNP Record

NCBI SNP ID: rs695871

## General

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## NCBI Resource Links

GenBank: [U70323](#)

LocusLink: no link established

Integrated Maps: *under construction*

## Submitter records for this ID

## Search

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Assay ID    Handle | Local Submitter ID

[ss869704](#)    FGU-CBT|SKB.2K.1.1

Release Date

Aug 2 2000 2:53PM

## Variation Summary

Assay sample size (number of chromosomes) : 430  
 Population data sample size (number of chromosomes) :  
 Total number of populations with frequency data: 1  
 Total number of individuals with genotype data: 0  
 Average estimated heterozygosity: 0.414  
 Average Allele Frequency:

C:	0.293
G:	0.707

## Validation Summary

Marker displays Mendelian segregation: UNKNOWN  
 PCR results confirmed in multiple reactions: YES  
 Homozygotes detected in individual genotype data: UNKNOWN  
 Insufficient genotype data to compute the goodness of fit to Hardy-Weinberg  
 Insufficient data to compute individual x genotype consistency measures  
 Validation status: *under construction*



## Submitter Contact Details

### General

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### Search

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[By Gene Name](#)

[Chromosome Report](#)

This batch's contact information:

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 email: shwetachoudhry@hotmail.com  
 lab: Functional Genomics Unit  
 institution: Centre for Biochemical Technology (CSIR)  
 address: Delhi University Campus, Mall Road, Delhi- 110007, India.

Handle information for the lab:

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 fax: +91-11-7257471  
 phone: +91-11-7416489  
 email: skb@cbt.res.in  
 lab: Functional Genomics Unit  
 institution: Centre for Biochemical Technology (CSIR)  
 address: Delhi University Campus, Mall Road, Delhi- 110007, India.



## SNP Population Details - 558

### General

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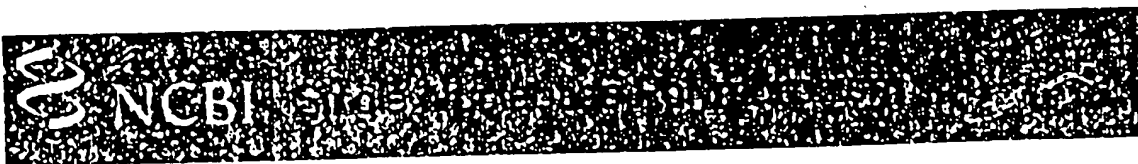
Submitter Population Handle: FGU-CBT  
Submitter Population ID: INDPOP  
Population Text:

Continent: Asia  
Nation: India

[View SNP used on this population](#)

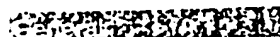
### Search

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Method

## SNP Method Details - 564

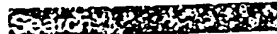

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[FTP SERVER](#)
[Database Schema](#)
[Blast SNP](#)
[Submission Form](#)

Submitter Method Handle: FGU-CBT  
 Submitter Method ID: SCA2-SNP  
 Method Text:

(SEQ ID NO: 1)

(SEQ ID NO: 2)

The region containing the SNP was PCR amplified using the primers SCA2-FP3 (5' CTCCGCCCTCAGACTGTTTGGTAG 3') and SCA2-RP3 (5' GTGGCCGAGGACGAGGAGAC 3'). Approximately 100ng of genomic DNA was amplified in a 50 µl reaction volume containing a final concentration of 5mM Tris, 25mM KCl, 0.75mM MgCl<sub>2</sub>, 0.05% gelatin, 20pmol of each primer and 0.5U of Taq DNA polymerase. Samples were denatured at 94°C for 3 min followed by 35 cycles of denaturation (94°C, 45sec), annealing (52°C, 30sec), extension (72°C, 45sec) and a final extension of 7 min at 72°C in a Perkin Elmer GeneAmp PCR System 9600. The PCR product was purified from band cut out of the agarose gel using QIAquick gel extraction kit (Qiagen) and was directly sequenced using dye terminator chemistry on an ABI Prism 377 automated DNA sequencer with the PCR primers.

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## Publication

## SNP Publication Details

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Submitter Handle: FGU-CBT

pmid:

MEDUID:

TITLE:

Single Nucleotide Polymorphism in SCA2 Gene.

AUTHOR:

CHOUDHRY, S.; BRAHMACHARI, S.K.

JOURNAL:

VOLUME:

SUPPL:

ISSUE:

I\_SUPPL:

PAGES:

YEAR: 2000

STATUS: 1- unpulished

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Search PubMed by author:

CHOUDHRY, S.; BRAHMACHARI, S.K.

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SNP Record

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[Method](#)
[Population](#)
[Publication](#)
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[Chromosome Report](#)

SNP:  
 Handle|local\_snp\_id: FGU-CBT | SKB.2K.1.2  
 NCBI Assay Id(ss#): 869705  
 Reference SNP Id(rs#): **696872**

STS Information: Not submitted

### From SNP Database:

Submitter Handle: **FGU-CBT**  
 Submitter Batch ID: **SKB.2K.1**  
 Release Date: **Aug 2 2000 2:53PM**  
 Molecular type: **Genomic**  
 No. of Chromosomes sampled: **215**  
 Synonym defined:  
 Organism: **Homo sapiens**  
 Population: **INDROF**  
 Submitter Method ID: **SCA2-SNP**  
 Citation:  
 Single Nucleotide Polymorphism in SCA2 Gene.

[View citation details](#)

NCBI Assay ID: 869705  
 Submitter SNP ID: SKB.2K.1.2  
 Synonyms:  
 LOCUSID: **6311**  
 Submitter STS ID:  
 STS Accession: not available  
 GenBank Accession: **U70323**  
 Gene Name: Human ataxin-2 gene/ Spinocerebellar ataxia 2 (SCA2) gene  
 Length: 459

### Flanking Sequence Information:

5' Assay: CTCGCCCTCA GACTGTTTGT GTAGCAACGG CAACGGCGGC GGCGCGTTTC GGCCCGGCTC  
 CCGGCGGGCTC CTTGGTCTCG GCGGGCCTCC CCGCCCCCTC GTCGTCGTCC TTCTCCCCCT  
 CGCCAGCCCG GGCGCCCTC CCGCCGCGCC AACCCGCGCC TCCCCGCTCG GCGCCCG (SEQ ID NO: 17)

Observed: T/C

3' Assay: GCGTCCCCGC CGCGTTCCGG CGTCTCCTTG GCGCGCCCGG CTCCCGGCTG TCCCCGCCCC  
 GCGTGCGAGC CGGTGTATGG GCCCTCACC ATGTGCT (SEQ ID NO: 18)

3' Flank: GAAGCCCCAG CAGCAGCAGC AGCAGCAGCA GCAACAGCAG CAGCAGCAAC AGCAGCAGCA  
 GCAGCAGCAG CAGCCGCGGC CCGCGGCTCC CAATGTCCGC AAGCCCGCGC GCAGCGGCCT  
 TCTAGCGTCG CCCGCGCGCG CGCCTTCGCC GTCTTCGTCC TCGGTCCTCT CGTCTCTGGC  
 CAC (SEQ ID NO: 16)

### Allele Frequency Information:

POP Batch Id: SKB.2K.1:

Submitter Handle: **FGU-CBT**  
 Submitter Method ID: **SCA2-SNP**  
 Citation:  
 Single Nucleotide Polymorphism in SCA2 Gene.

Handle|PopulationID: FGU-CBT|INDROF  
 No. of Chromosomes Sampled: 215  
 Allele: C = 0.293 / T = 0.707





RefSNP Record

## Reference SNP Record

NCBI SNP ID: rs695872

### NCBI Resource Links

GenBank: [U70323](#)

LocusLink: no link established

Integrated Maps: *under construction*

### Submitter records for this ID:

Assay ID	Handle   Local Submitter ID	Release Date
<a href="#">ss869705</a>	FGU-C8T SKB.2K.1.2	Aug 2 2000 2:53PM

### Variation Summary:

Assay sample size (number of chromosomes) : 430  
 Population data sample size (number of chromosomes) :  
 Total number of populations with frequency data: 1  
 Total number of individuals with genotype data: 0  
 Average estimated heterozygosity: 0.414  
 Average Allele Frequency:

C:	0.293
T:	0.707

### Validation Summary:

Marker displays Mendelian segregation: UNKNOWN  
 PCR results confirmed in multiple reactions: YES  
 Homozygotes detected in individual genotype data: UNKNOWN  
 Insufficient genotype data to compute the goodness of fit to Hardy-Weinberg  
 Insufficient data to compute individual x genotype consistency measures  
 Validation status: *under construction*

Publication

## SNP Publication Details

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[FAQ](#)  
[RefSNP Summary Info](#)  
[FTP SERVER](#)  
[Database Schema](#)  
[Blast SNP](#)  
[Submission Form](#)

Submitter Handle: FGU-CBT  
 pmid:  
 MEDID:  
 TITLE:  
 Single Nucleotide Polymorphism in SCA2 Gene.  
 AUTHOR:  
 CHOUDHRY,S.; BRAHMACHARI,S.K.  
 JOURNAL:  
 VOLUME:  
 SUPPL:  
 ISSUE:  
 I\_SUPPL:  
 PAGES:  
 YEAR: 2000  
 STATUS: 1- unpublished

Search PubMed by author:  
CHOUDHRY,S.; BRAHMACHARI,S.K.

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Population

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[FTP SERVER](#)

[Database Schema](#)

[Blast SNP](#)

[Submission Form](#)

Submitter Population Handle: FGU-CBT

Submitter Population ID: INDPOP

Population Text:

Continent: Asia

Nation: India

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## SNP Method Details - 564

Method

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Submitter Method Handle: FGU-CBT

Submitter Method ID: SCA2-SNP

Method Text:

(SEQ ID NO:1)

The region containing the SNP was PCR amplified using the primers SCA2-FP3 (5' CTCGCCCTCAGACTGTTTGGTAG 3') and SCA2-RP3 (5' GTCGCCGAGGACGAGGAGAC 3'). Approximately 100ng of genomic DNA was amplified in a 50 ml reaction volume containing a final concentration of 5mM Tris, 25mM KCl, 0.75mM MgCl<sub>2</sub>, 0.05% gelatin, 20pmol of each primer and 0.5U of Taq DNA polymerase. Samples were denatured at 94°C for 3 min followed by 35 cycles of denaturation (94°C, 45sec), annealing (52°C, 30sec), extension (72°C, 45sec) and a final extension of 7 min at 72°C in a Perkin Elmer GeneAmp PCR System 9600. The PCR product was purified from band cut out of the agarose gel using QIAquick gel extraction kit (Qiagen) and was directly sequenced using dye terminator chemistry on an ABI Prism 377 automated DNA sequencer with the PCR primers.

(SEQ ID NO:2)

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Submitter

## Submitter Contact Details

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This batch's contact information:

handle: FGU-CBT  
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 lab: Functional Genomics Unit  
 institution: Centre for Biochemical Technology (CSIR)  
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---

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 phone: +91-11-7416489  
 email: skb@cbt.res.in  
 lab: Functional Genomics Unit  
 institution: Centre for Biochemical Technology (CSIR),  
 address: Delhi University Campus, Mall Road, Delhi- 110007, India.



# Nucleotide

Search  for

☐ Hide Brief and LinkBar

☐ 1: GI = "1679683" [GenBank] Human ataxin-2 (SCA2) mRNA.... PubMed, Protein, Related Sequences, Taxonomy, OMIM, LinkOut

LOCUS HSU70323 4481 bp mRNA PRI 20-NOV-1996  
 DEFINITION Human ataxin-2 (SCA2) mRNA, complete cds.  
 ACCESSION U70323  
 VERSION U70323.1 GI:1679683  
 KEYWORDS  
 SOURCE human.  
 ORGANISM Homo sapiens  
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;  
 Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.  
 REFERENCE 1 (bases 1 to 4481)  
 AUTHORS Pulst, S.-M., Nechiporuk, A., Nechiporuk, T., Gispert, S., Chen, X.-N.,  
 Lopes-Cendes, I., Pearlman, S., Starkman, S., Orozco-Diaz, G.,  
 Lunkes, A., DeJong, P., Rouleau, G.A., Auburger, G., Korenberg, J.R.,  
 Figueroa, C. and Sahba, S.  
 TITLE Moderate expansion of a normally biallelic trinucleotide repeat in  
 spinocerebellar ataxia type 2  
 JOURNAL Nature Genet. 14 (3), 269-276 (1996)  
 MEDLINE 97051920  
 REFERENCE 2 (bases 1 to 4481)  
 AUTHORS Pulst, S.-M.  
 TITLE Direct Submission  
 JOURNAL Submitted (10-SEP-1996) Medicine, Cedars-Sinai, 8700 Beverly Blvd.,  
 Los Angeles, CA 90048, USA  
 FEATURES  
 source Location/Qualifiers  
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 MSLKPOQQQQQQQQQQQQQQQQQQQQQQPPAAANVRKPGGSGLLASPAAPSPSSSV  
 SSSSATAPSSVVAATSCGGRPGLGRNSNKLPGSTISFDGIYANKRMVHILTSVVG  
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BASE COUNT 1144 a 1380 c 1014 g 943 t

file:///Macintosh%20HD/Desktop%20of%20older/abn/seq/Show%20%20SCA2%20nucleotide

Figure : 6

Wednesday, September 27, 2000

NCBI Sequence Viewer

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## ORIGIN

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1201  gcaaaaagag atgcttttcc tgactctgct atcagtgtca aagtgaatgg cgaacacaaa
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1321  ttggaatatg acgtatctaa tggatgggat cccaatgata tgtttcgata taatgaagaa
1381  aattatgggt tagtgtctac gtatgatagc agtttatctt cgtatcacgt gcccttagaa
1441  agagataaact cagaagaact tttaaaacgg gaagcaaggg caaacagtt agcagaagaa
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1981  gctctctacta tgcctaaacg catgtcttca gaagggcctc caaggtatgc cccaaggccc
2041  cagcgacatc ctgcaaatca cagagtttct gctgggaggg gttccatata cagtggccta
2101  gaatttgtat cccacaaccc acccagtga aagcgtactc ctccagttag aaggaccagt
2161  ccctcggggg gaacgtgttc atcagtgttc agtgggggtc caagattatc cccataaact
2221  catagaccac ggtctccccc acagaacagt attggaataa ccccagtggt gccagtctct
2281  gcttctcccc aagctgttat tattccaact gaagctgttg ccatgcctat tccagctgca
2341  tctcctacgc ctgctagttc tgcctcgaac agagctgtta ccccttctag tgaggctaaa
2401  gattccaggg ttcaagatca gaggcagaac tctcctcgag ggaataaaga aaatatataa
2461  cccaatgaaa catcacatag cttctcaaaa gctgaanaaa aaggtatata accagtgtgt
2521  tctgaacata gaaaacagat tgatgattta aagaaattta agaattatgt taggttacag
2581  ccaagtctta cttctgaatc tatggatcaa ctactaaaca aaatatagaa gggagaaaaa
2641  tcaagagatt tcatcaagaa caaanttgaa ccaagtgtca aggtattctt cattgaaaaa
2701  agcagcagca actgtaccag tggcagcagc aagccgaata gccccagcat ttccccctca
2761  atacttagta acacggagca caagagggga cctgaggtca cttcccaagg ggttcagact
2821  tccagccccc catgtaaaca agagaaagac gataaggaag agaagaaaga cgcagctgag
2881  caagttagga aatcaacatt gaatcccaat gcaaaaggag tcaacccacg tctctctct
2941  cagccaaaag cttctactac cccaacttca cctcggcctc aagcacaacc tagcccatct
3001  atggtgtggc atcaaacgcc aactccagtt tatactcagc ctgtttgttt tgcacaaaat
3061  atgatgtatc cagtcctcag gagcccgagg gtgcaacctt tatacccaat acctatgacg
3121  cccatgccag tgaatcaagc caagacatat agagcagtag caaatatgcc ccaacagcgg
3181  caagaccagc atcatcagag tggcatgatg caccagcggt cagcagcggg cccaccgatt
3241  gcagccaccc caccagctta ctccacgcaa tatgttgctt acagtcttca gcagtctcca
3301  aatcagcccc ttgttcagca tgtgccacat tatcagttct agcatcttca tgtctatagt
3361  cctgtaatac agggtaatgc tagaatgatg gcaccaccaa cacacgcccc gcttggttta
3421  gtatctctct cagcaactca gtacggggct catgagcaga cgcagtcgat gtatgcattg
3481  cccaaattac catacaacaa ggagacaagc ccttcttctt actttgccaat ttccacgggc
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3601  cagccttcag ctacccccac tggacagcag caaagccaac atggtggaag tcatcctgca
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3781  gcctccaaca cgcagtcgcc acagaatagt ttcccagcag caaacagac tgtctttacg
3841  atccatctct ctccagttca gccggtgtat accaaccac cccacatggc ccacgtacct
3901  caggtctcat tacagtccag aatgttctct tctcatccaa ctgcccattg gccaatgatg
3961  ctaagtacga cacagccacc gcggtgtccc caggccggcc tcgctcaaa gtcactacag
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4321  tatgctgttt cagagtcctc caggtacccc agctctgctt gccgaactg gaagttatct
4381  attttttaat aacccttgaa agtcatgaac acatcagcta gcaaaagaag taacaagagt
4441  gattcttgct gctattactg ctaaaaaaa aaaaaaaa a

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(SEQ ID NO: 20)

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Fig.6(Cont.)